

54. A genetic marker set comprising three or more single nucleotide polymorphisms (SNPs), wherein each allele of the SNPs has an allelic frequency between 0.001 and 0.999.

55. A genetic marker set according to claim 54, wherein each allele of the SNPs has an allelic frequency between 0.01 and 0.99.

56. A method for preparing a genetic marker set from a genome of a species of interest, comprising:

selecting a set of three or more single nucleotide polymorphisms (SNPs) in the genome of the species of interest,

wherein each allele of the SNPs has an allelic frequency between 0.001 and 0.999.

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57. A method according to claim 56, wherein each allele of the SNPs has an allelic frequency between 0.01 and 0.99.

58. A method according to claim 56, wherein the SNPs are bi-allelic.

59. A method according to claim 56, wherein the SNPs are tri-allelic.

60. A method according to claim 56, wherein the SNPs are tetra-allelic.

61. A method according to claim 56, wherein the SNPs arise from insertions or deletions in the genome of the species of interest.

62. A method according to claim 56, wherein the SNPs are selected from the group consisting of bi-allelic SNPs, tri-allelic SNPs, tetra-allelic SNPs and any combination thereof.

63. A method according to claim 62, wherein at least one of the SNPs arise from insertions or deletions in the genome of the species of interest.

64. A method according to claim 56, wherein the species of interest is selected from the group consisting of animals, plants, fungi, yeast and *C. elegans*.

65. A method according to claim 64, wherein the species of interest is a mammal.

66. A method according to claim 65, wherein the mammal is selected from the group consisting of human, non-human primates, dogs, cats, cattle, sheep, horses, mouse, rat and rabbit.

67. A method according to claim 66, wherein the species of interest is human.

68. A method according to claim 66, wherein the species of interest is horse.

69. A method according to claim 64, wherein the species of interest is selected from the group consisting of corn, wheat, soy, peas and rice.

70. A method according to claim 56, wherein the genome comprises RNA.

71. A method for genotyping an individual from a species of interest, comprising:  
(a) selecting a set of genetic markers, in the genome of the species of interest, comprising three or more single nucleotide polymorphisms (SNPs), wherein each allele has an allelic frequency between 0.001 and 0.999, and  
(b) determining the base identity at each polymorphic site for the individual.

72. A method according to claim 71, wherein each allele has an allelic frequency between 0.01 and 0.99.

73. A method for determining the number of matching SNP alleles between DNA from a target individual and DNA from a reference individual of the same species as the target individual, comprising:

determining, for one or more single nucleotide polymorphisms of the target individual, and for one or more corresponding single nucleotide polymorphisms of the reference individual, whether the polymorphisms contain the same single nucleotide at their respective corresponding polymorphic sites, and thus determining the number of matching SNP alleles between the DNA from the target individual and DNA from the reference individual.

74. A method according to claim 73, wherein the determining of matching SNP alleles is sufficient to establish that the reference individual is not a parent of the target individual.

75. A method according to claim 73, wherein the reference individual has a trait linked to the polymorphism, and the determining of matching SNP alleles is sufficient to predict whether the target individual also has the trait.

76. A method according to claim 73, wherein the reference individual has a first and second trait, and the determining of matching SNP alleles is sufficient to establish a genetic linkage between the traits.

77. A method according to claim 73, wherein the determining whether the polymorphisms contain the same single nucleotide at there respective corresponding polymorphic sites is accomplished by a method comprising:

(a) incubating a sample of nucleic acid comprising the single nucleotide polymorphism of the target individual, or the single nucleotide polymorphism of the reference individual, in the presence of a nucleic acid primer and at least one chain terminating nucleotide, derivative thereof, or chain terminating nucleotide analog, under conditions sufficient to permit a polymerase mediated, template-dependent extension of the primer, the extension causing the incorporation of a single

dideoxynucleotide to the 3'-terminus of the primer, the single dideoxynucleotide being complementary to the single nucleotide of the polymorphic site of the polymorphism;

(b) permitting the template-dependent extension of the primer molecule, and the incorporation of the single dideoxynucleotide; and

(c) determining the identity of the nucleotide incorporated into the polymorphic site, the identified nucleotide being complimentary to the nucleotide of the polymorphic site.

78. A method according to claim 77, wherein in step (a), the primer is immobilized to a solid support, and wherein in step (b), the template-dependent extension of the primer is conducted on the immobilized primer.

79. A method according to claim 77, wherein, in step (a), the sample is processed to amplify a nucleic acid containing the polymorphism prior to the incubation.

80. A method according to claim 77, wherein step (a) additionally includes using a non-invasive swab to collect the sample of DNA from the target individual.

81. A method according to claim 77, wherein in step (a), the polymerase mediated, template-dependent extension of the primer is conducted in the presence of at least two dideoxynucleotide triphosphate derivatives selected from the group consisting of ddATP, ddTTP, ddCTP and ddGTP, but in the absence of dATP, dTTP, dCTP and dGTP.

82. A method of establishing the genetic similarity of two or more individuals from a species of interest, comprising:

(a) selecting a set of genetic markers from the species of interest comprising three or more single nucleotide polymorphisms (SNPs) wherein each allele has a frequency between 0.001 and 0.999;

(b) determining the base identity at each polymorphic site for each individual, and

(c) determining the number of matching polymorphisms present in any pair of individuals, thus establishing the genetic similarity of the two or more individuals from the species of interest.

83. A method for determining the probability that an unknown sample of nucleic acid molecules is derived from a known individual, comprising:

(a) selecting a reference set of genetic markers comprising three or more single nucleotide polymorphisms (SNPs) having a cumulative probability of identity of greater than about 0.8;

(b) identifying alleles present at each SNP site of the reference set of genetic markers in the known individual by determining the base identity at each SNP site and identifying alleles present at each SNP site of the reference set of genetic markers in the unknown sample of nucleic acid molecules by determining the base identity at each SNP site for the unknown sample of nucleic acid molecules;

(c) determining the matches between the SNP sites of the individual and the unknown sample of nucleic acid molecules, and

(d) calculating the probability of identity from the matches determined in step (c) and the allelic frequency of the SNPs in the matching genetic markers, and thus determining the probability that the unknown sample of nucleic acid molecules is derived from the known individual.

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84. A method according to claim 83, wherein the cumulative probability of identity is greater than about 0.95.

85. A method according to claim 84, wherein the cumulative probability of identity is greater than about 0.99.

86. A method according to claim 83, wherein the nucleic acid molecules are DNA.

87. A method for excluding the possibility that an unknown sample of nucleic acid molecules are derived from a known individual, comprising:

(a) selecting a reference set of genetic markers comprising three or more single nucleotide polymorphisms (SNPs) having a cumulative probability of exclusion of greater than about 0.8;

(b) determining the base identity at each polymorphic site in the unknown sample and determining the base identity at each polymorphic site in the sample derived from the known individual;

(c) determining the number of matches, and

(d) calculating a probability of exclusion from the matches determined in (c) and the allelic frequencies of the SNPs in the reference set.

88. A method according to claim 87, wherein the probability of exclusion is greater than about 0.95.

89. A method according to claim 87, wherein the probability of exclusion is greater than about 0.99.

90. A method according to claim 87, wherein the nucleic acid molecules are DNA.

91. A method of excluding the possibility that an individual is the progeny of a possible ancestor, comprising:

(a) selecting a set of genetic markers comprising three or more single nucleotide polymorphisms (SNPs) having a cumulative probability of exclusion of greater than 0.8;

(b) determining the base identity at each polymorphic site for the individual and the possible ancestor;

(c) determining matches between the polymorphic sites of the individual and the possible ancestor;

(d) calculating from the matches obtained in (c) and the allelic frequencies of the SNPs in the set of genetic markers the probability that the individual is not the progeny of the possible ancestor.

92. A method according to claim 91, wherein the probability of exclusion is greater than 0.95.

93. A method according to claim 92, wherein the probability of exclusion is greater than 0.99.

94. A method of generating a genetic map of an individual, comprising:

(a) providing a single nucleotide polymorphism (SNP) marker set which comprises three or more single nucleotide polymorphisms (SNPs), wherein each allele has an allelic frequencies between 0.001 and 0.999;

(b) identifying the alleles present in an ancestor of the individual by determining the base identity at each SNP site of the ancestor of the individual and identifying the alleles present in the individual by determining the base identity at each SNP site of the individual;

(c) determining the number of matches between the individual and the ancestor;

(d) calculating the extent of genetic linkage between each allele from the number of matches of step (c) and the probability that any pair of alleles found in the individual were inherited from the same ancestor based on the allelic frequencies in the reference marker set, thereby generating the genetic map of the individual.

95. A method according to claim 94, wherein the SNP sites used to construct the genetic map are randomly distributed throughout the genome of the species.

96. A method according to claim 94, wherein the SNP sites used to construct the genetic map are distributed throughout a single chromosome.

97. A method according to claim 94, wherein the SNP sites used to construct the genetic map are distributed throughout a sequence of less than about 10 MB.

98. A method according to claim 94, wherein the ancestor is selected from the group consisting of parent and grandparent.

99. A method for determining the probability that a target individual will have a particular trait, comprising:

(a) identifying a single nucleotide present at a polymorphic site of a single nucleotide polymorphism, wherein the single nucleotide is present in more than 51% of a set of reference individuals;

(b) determining whether a single nucleotide present at a polymorphic site of a corresponding single nucleotide polymorphism of the target individual has the same identity as the single nucleotide present at the polymorphic site of the 51% of reference individuals exhibiting the trait, and

(c) determining the probability that the target individual of step (b) will have the particular trait.

100. A method according to claim 99, wherein the trait is a genetic disease.

101. A method of associating the presence of a particular trait of interest found in an individual with a particular allele found at a SNP site, comprising:

(a) selecting two or more SNP sites wherein each allele has an allelic frequency between 0.001 and 0.999;

(b) identifying the alleles present in one or more individuals having the trait of interest by determining the base identity at each SNP site, and

(c) determining whether one or more alleles are present in the population of individuals having the trait of interest at a frequency greater than the allelic frequency, thus associating the alleles with the trait of interest.

102. A method according to claim 101, wherein each allele has an allelic frequency between 0.01 and 0.99.

103. A method according to claim 101, wherein the trait of interest is a predisposition to a genetic disease.

104. A method according to claim 101, wherein the trait of interest is a genetic disease.

105. A method according to claim 101, wherein the trait of interest is a response to a drug.

106. A method according to claim 105, wherein the response to a drug is an adverse drug reaction.

107. A method according to claims 83, 91, 94, 99 or 101, wherein the individual or target individual is selected from the group consisting of animals, plants, fungi, yeast and *C. elegans*.

108. A method according to claim 107, wherein the individual or the target individual is a mammal.

109. A method according to claim 108, wherein the mammal is selected from the group consisting of human, non-human primates, dogs, cats, cattle, sheep, horses, mouse, rat and rabbit.

110. A method according to claim 108, wherein the individual or target individual is a human.

111. A method according to claim 108, wherein the individual or target individual is a horse.

112. A method according to claim 107, wherein the individual or target individual is selected from the group consisting of corn, wheat, soy, peas and rice.

113. A method for identifying single nucleotide polymorphic sites in a genome of a species of interest, comprising:

(a) isolating a plurality of DNA fragments from the genome of a population of individual representatives of the species of interest, wherein each fragment corresponds to a location of the genome and the fragments are between about 0.1 kb and 10.0 kb;

(b) sequencing the fragments of DNA to determine the nucleotide sequences of each fragment, and

(c) comparing the sequence of each fragment to corresponding fragments from other individual representatives of the species of interest to identify sites of sequence variation, thereby determining the alleles present in the location of the genome.

114. A method according to claim 113, wherein the plurality of DNA fragments is from a population of between 10 to 100 individual representatives of the same species.  
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115. A method according to claim 113, wherein the plurality of DNA fragments is from a population of between 100 to 1000 individual representatives of the same species.

116. A method according to claim 113, wherein the plurality of DNA fragments is from a population of between 100 and 10,000 individual representatives of the same species.

117. A method according to claim 113, wherein the fragments are between 0.5 kb and 3.0 kb.

118. A method according to claim 113, wherein the fragments are sequenced by dideoxy sequencing.

119. A method according to claim 113, wherein the fragments are isolated by amplification using oligonucleotide primers.

120. A method for determining allelic frequency at a single nucleotide polymorphic site, comprising:

- (a) isolating a plurality of DNA fragments from a population of two or more individual representatives of a species of interest, wherein each fragment corresponds to a location of the genome and the fragments are between about 0.1 kb and 10.0 kb;
- (b) sequencing the fragments of DNA to determine the nucleotide sequences of each fragment;
- (c) comparing the sequence of each fragment to corresponding DNA fragments from different individual representatives of the species of interest and identifying single nucleotide polymorphic sites having at least two alleles,
- (d) determining the base identity of each allele present in the location of the genome, and
- (e) calculating the allelic frequency for each allele by dividing the frequency at which each allele appears in the sample set by the total number of individuals.

121. A method according to claim 120, wherein the plurality of DNA fragments is from a population of between 10 to 100 individual representatives of the same species.

122. A method according to claim 120, wherein the plurality of DNA fragments is from a population of between 100 to 1000 individual representatives of the same species.

123. A method according to claim 120, wherein the plurality of DNA fragments is from a population of between 100 and 10,000 individual representatives of the same species.